

REACH  **24**

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7TH INTERNATIONAL
**LFS ASSOCIATION
SYMPOSIUM**

HOSTED BY



Penn Medicine



Children's Hospital
of Philadelphia

LIFE

LI-Fraumeni Exploration Consortium

WELCOME!

Program

October 19–22, 2024

7th International LFS Association Symposium

Hosted by Penn Medicine/CHOP and in partnership with the LiFE Consortium

Program Agenda

Friday, October 18th

3.00pm – 6.00pm
6.00pm – 8.00pm

| **The Inn at Penn**

3600 Sansom St, Philadelphia, PA, 19104

Registration and Check-in – The Inn at Penn Lobby
Welcome Cocktail Reception – The Inn at Penn

Saturday, October 19th

| **Penn Smilow Center for Translational Research**

3400 Civic Center Blvd, Philadelphia, PA, 19104

7.00am – 8.30am

Registration

7.00am – 8.30am

Breakfast

8.30am – 10.00am

Welcome

2024 LFS Symposium Planning Committee

Introduction

Jenn Perry

Introduction by Abramson Cancer Center

Katherine Nathanson, MD

Theirry Frebourg Memorial Lecture

Progress and Promise in Li Fraumeni Syndrome: A Collaborative View
Judy Garber, MD, MPH

10.00am – 10.20am

Break

10.20am – 11.30am

Low Risk Attenuated LFS

Session Chair: Kara N. Maxwell, MD, PhD

Co-Chair: Christian Kratz, MD

10.20am – 10.35am

The genetics of tumor suppression by p53: what can we learn from hypomorphic p53 variants?

Maureen Murphy, PhD

10.35am – 10.50am

Modifiers of Cancer Risk in TP53 p.R337H Variant Carriers: The Role of Genetic and Environmental factors

Emilia Pinto, PhD

10.50am – 11.05am

Cancer Histories of Families with Hypomorphic TP53 variants and Implications for Management

Huma Rana, MD, MPH

11.05am – 11.20am	Genetic counseling perspectives on reduced penetrance/attenuated Li-Fraumeni Syndrome Jacquelyn Powers, MS, CGC
11.20am – 11.30am	<i>Q & A session</i>
11.30am – 12.30pm	Genotype–Phenotype and Clinical Genetics Studies Session 1 Session Chair: Phuong Mai, MD, MS Co-Chair: Dana Farengo-Clark, MS, CGC
11.30am – 11.045am	Estimating Penetrance from the LiFT UP Study Stephen Gruber, MD, PhD, MPH
11.45am – 12.00pm	Outcomes and clinical phenotypes of LFS breast cancer Renata Sandoval, MD, PhD
12.00pm – 12.15pm	Distinct genomic and immunologic tumor evolution in germline <i>TP53</i> -driven breast cancers Kara N. Maxwell, MD, PhD
12.15pm – 12.30pm	<i>Q & A session</i>
12.30pm – 1.30pm	Lunch
1.30pm – 2.15pm	LFSA International Youth Panel
2.15pm – 3.00pm	LFSA Programming <i>Patient Story with Debora Weiss</i>
3.00pm – 3.30pm	Break
3.30pm – 5.00pm	Parents' Guide to LFS Session Chair: Allison Werner-Lin, PhD, LCSW Co-Chair: Suzanne MacFarland, MD
3.30pm – 3.45pm	Talking with children and Li-Fraumeni Syndrome Allison Werner-Lin, PhD, LCSW
3.45pm – 4.00pm	Transition from pediatric to adult care - the Karolinska model Meis Omran, MD, PhD
4.00pm – 4.15pm	Supporting Adolescents and Young Adults with LFS and other Cancer Risk Syndromes: the AYA-RISE Study Junne Kamihara, MD, PhD
4.15pm – 4.30pm	Supporting Parents and Preparing Teens to Manage LFS Care Kristin Zelle, MS, CGC
4.30pm – 5.00pm	<i>Q & A session</i>
5.00pm – 7.00pm	Poster Session and Reception Penn Smilow Center for Translational Research 3400 Civic Center Blvd., Philadelphia, PA, 19104

Sunday, October 20th

| CHOP Hub for Clinical Collaboration

3500 Civic Center Blvd, Philadelphia, PA, 19104

7.30am – 8.30am

Breakfast

8.30am – 9.10am

Welcome and Introduction

Garrett Brodeur, MD

Joseph Fraumeni Honorary Lecture

Li-Fraumeni Syndrome: Past Hopes and Future Promise

David Malkin, MD

9.10am – 10.25am

TP53 Variant Interpretation

Session Chair: Suzanne MacFarland, MD

Co-Chair: Kara Maxwell, MD, PhD

9.10am – 9.25am

Suspected clonal hematopoiesis as a natural functional assay of *TP53* germline variant pathogenicity

Cristina Fortuno, PhD

9.25am – 9.40am

ClinGen *TP53* Variant Curation Expert Panel v2 Specifications Improve Analysis of *TP53* Variants

Megan Frone, MS, CGC

9.40am – 9.55am

Tumor Spectrum and Temporal Cancer Trends in Adult Carriers of Li-Fraumeni Syndrome: Implications for Personalized Screening Strategies in *TP53* R337H Carriers

Maria Isabel Achatz, MD, PhD

9.55am – 10.10am

Mechanisms of incomplete penetrance of germline *TP53* variants

Pierre Hainaut, PhD

10.10am – 10.25am

Deep mutational scanning supports a multistage model of mutant *TP53*-driven tumorigenesis

Andrew Giacomelli, PhD

10.25am – 10.40am

Q & A session

10.40am – 10.55am

Break

10.55am – 12.45pm

Genotype-Phenotype and Clinical Genetics Studies Session 2

Session Chair: Banu Arun, MD

Co-Chair: Jacquelyn Powers, MS, CGC

10.55am – 11.10am

The Li-Fraumeni Syndrome Registry in Germany

Christian Kratz, MD

11.10am – 11.25am

Li-Fraumeni Syndrome in the UK; Manchester experience

Emma Woodward, FRCP, PhD

11.25am – 11.40am	Updates from the French LFS experience Gaëlle Bougeard, PhD	
11.40am – 11.55am	Cancer risk modeling for deleterious mutations in <i>TP53</i> using a multi-center consortium Wenyi Wang, PhD	
11.55am – 12.10pm	Risk of therapy-related subsequent cancers in individuals with germline <i>TP53</i> variants Anita Villani, MD, MSc	
12.10pm – 12.25pm	Identification of <i>TP53</i> germline variants in pediatric patients undergoing tumor testing: strategies, prevalence, and challenges" Minjie Luo, PhD	
12.25pm – 12.40pm	Q & A session	
12.40pm – 1.15pm	Lunch	
1.15pm – 3.00pm	Breakout Sessions	
1.15pm – 2.00pm	<p><i>Session 1A: Breast Cancer in LFS – Before and After (Hybrid)</i></p> <p><u>Leads:</u> Bita Nehoray, MS, CGC Jacquelyn Powers, MS, CGC</p> <p><u>Expert Panel:</u> Christine Edmond, MD Judy Garber, MD, MPH Alina Mateo, MD, MS, FACS Camella J. Rising, PhD, MS, RDN</p>	<p><i>Session 1B: Parents’ guide to the LFS Universe (Hybrid)</i></p> <p><u>Leads:</u> Allison Werner-Lin, PhD, LCSW Bailey Gallinger, MSc, CGC</p> <p><u>Expert Panel:</u> Suzanne MacFarland, MD Kristin Zellej, MS, CGC</p>
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3.10pm – 3.25pm	Break	
3.25pm – 3.45pm	LFSA Programming “Your Family is My Family” with author Lawrence Ingrassia	

3.45pm – 4.15pm

Remarks by LFSA Medical Director

Robert Lufkin, DO

4.15pm – 5.30pm

Health Behavior and Medical Decision Making

Session Chair: Rowan Forbes Shepherd, PhD

Co-Chair: Kristin Zelle, MS, CGC

4.15pm – 4.30pm

Psychosocial correlates of reported physical activity among adults with LFS and their family members from a National Cancer Institute cohort

Camella J. Rising, PhD, MS, RDN

4.30pm – 4.45pm

Implementation of a support group program for individuals and families impacted by Li-Fraumeni Syndrome: A pilot study

Kelsey Breen, MS, CGC

4.45pm – 5.00pm

Adolescent and Young Adult (AYA) Management of Cancer Risk During the Transition to Adulthood

Lisa Schwartz, PhD

5.00pm – 5.15pm

Improving psychosocial support for families with Li-Fraumeni Syndrome Families: Insights from Germany

Antonia Rabe, PsyM

5.15pm – 5.30pm

Q & A session

6.30pm – 8.30pm

Open Evening with optional “Flavors of Philly” Light fare

The Inn at Penn Library

3600 Sansom St, Philadelphia, PA, 19104

Monday, October 21st

| CHOP Hub for Clinical Collaboration
3500 Civic Center Blvd, Philadelphia, PA

7.30am – 8.30am

Breakfast

8.30am – 9.30am

P53 Biology

Session Chair: David Malkin, MD

Co-Chair: Suzanne MacFarland, MD

8.30am – 8.50am

Testing the idea of cancer prevention for LFS
Arnold Levine, PhD

8.50am – 9.05am

TBA
Ran Kafri, PhD

9.05am – 9.20am

Zebrafish models of LFS provide new insights into disease progression and cancer prevention
Jason Berman, MD

9.20am – 9.30am

Q & A session

9.30am – 10.30am

P53 Biology and Mouse models

Session Chair: Sharon Savage, MD

Co-Chair: Kara Maxwell, MD, PhD

9.30am – 9.45am

The Complexity and Controversy of Mutant p53
Carol Prives, PhD

9.45am – 10.00am

In vivo mutant p53 activities
Guillermo Lozano, PhD

10.00am – 10.15am

Deconstructing mechanisms of p53-mediated lung cancer suppression
Laura Attardi, PhD

10.15am – 10.30am

Q & A session

10.30am – 10.50am

Break

10.50am – 12.00pm

Targeted Therapy

Session Chair: Joshua Schiffman, MD

Co-Chair: Payal Khincha, MBBS, MSHS

10.50am – 11.05am

Nonsense mutant *TP53* as a therapeutic target in LFS
Klas Wiman, MD, PhD

11.05am – 11.20am

Precision Targeting of p53 Mutant Cancers
Christine Eischen, PhD

11.20am – 11.35am

Can We Delay Cancer Development in Li-Fraumeni Syndrome Patients?
Kanaga Sabapathy, PhD

11.35am – 11.50am	Metformin as a pharmacopreventive agent in LFS (MILI): an update on the international efforts Payal Khincha, MBBS, MSHS		
11.50am – 12.00pm	Q & A session		
12.10pm – 1.00pm	Lunch		
1.00pm – 3.00pm	Breakout Sessions		
1.00pm – 1.45pm	<table border="0" style="width: 100%;"> <tr> <td style="width: 50%; vertical-align: top;"> <p>Session 2A: Spirit/Mind/Body (Hybrid)</p> <p><u>Leads:</u> Camella J. Rising, PhD, MS, RDN Rose McGee, MS, CGC</p> <p><u>Expert Panel:</u> Tracey Jubelirer, MD Tonia Kulp, MS, C-IAYT</p> </td> <td style="width: 50%; vertical-align: top;"> <p>Session 2B: Hot Topics in LFS Surveillance (Hybrid)</p> <p><u>Leads:</u> Alison Schwartz Levine, MS, CGC Rowan Forbes Shepherd, PhD</p> <p><u>Expert Panel:</u> Mark Rosen, MD, PhD Kara Maxwell, MD, PhD Lisa States, MD</p> </td> </tr> </table>	<p>Session 2A: Spirit/Mind/Body (Hybrid)</p> <p><u>Leads:</u> Camella J. Rising, PhD, MS, RDN Rose McGee, MS, CGC</p> <p><u>Expert Panel:</u> Tracey Jubelirer, MD Tonia Kulp, MS, C-IAYT</p>	<p>Session 2B: Hot Topics in LFS Surveillance (Hybrid)</p> <p><u>Leads:</u> Alison Schwartz Levine, MS, CGC Rowan Forbes Shepherd, PhD</p> <p><u>Expert Panel:</u> Mark Rosen, MD, PhD Kara Maxwell, MD, PhD Lisa States, MD</p>
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2.45pm – 3.00pm	Break		
3.00pm – 4.15pm	Barriers to care in LFS Session Chair: Michael Hall, MD, MS Co-Chair: Angela Bradbury, MD		
3.00pm – 3.15pm	Screening for cancer predisposition syndromes in children and adolescents with cancer - MIPOGG app Catherine Goudie, MD, MSc		
3.15pm – 3.30pm	Identification of Patients with LFS from Cancer Diagnosis in Kids and Adults Kristin Zelle, MS, CGC & Dana Farengo-Clark, MS, CGC		
3.30pm – 3.45pm	Health Equity and Li-Fraumeni Syndrome Haley Newman, MD, MSCE		
3.45pm – 4.00pm	Clinical care of the gender diverse patient with LFS Shelly McQuaid, MS, CGC		

4.00pm – 4.15pm

Q & A session

5.00pm – 7.00pm

International Dinner

The Inn at Penn

3600 Sansom St, Philadelphia, PA, 19104

Tuesday, October 22nd

| CHOP Hub for Clinical Collaboration

3500 Civic Center Blvd, Philadelphia, PA, 19104

7.00am – 8.00am

Hot Breakfast Buffet

8.00am – 9.00am

Early Detection and Screening: Radiology

Session Chair: Lisa J. States, MD

Co-Chair: Mark Rosen, MD, PhD

8.00am – 8.15am

Whole-Body MRI Screening in Infants, Children and Adolescents with LFS
Mary-Louise Greer, MBBS, FRANZCR

8.15am – 8.30am

Whole-body MRI in patients with LFS
Ashkan Malayeri, MD

8.30am – 8.45am

Gastrointestinal and pancreatic cancer screening in Li-Fraumeni Syndrome
Bryson Katona, MD, PhD

8.45am – 9.00am

Q & A session

9.00am – 10.00am

Early Detection and Screening – cfDNA

Session Chair: Payal Khincha, MBBS, MSHS

Co-Chair: Susan Domchek, MD

9.00am – 9.15am

Building a path to clinical validation of liquid biopsies for cancer screening in cancer risk syndromes
Brian D. Crompton, MD

9.15am – 9.30am

Liquid Biopsy in LFS: Moving Towards a Clinical Trial
Suzanne P. MacFarland, MD

9.30am – 9.45am

The CHARM of it all: Putting cell-free DNA into practice in a prospective trial
Trevor Pugh, PhD

9.45am – 10.00am

Q & A session

10.10am – 10.30am

Break

10.30am – 12.00pm

Panel Discussion

12.10pm – 1.00pm

Program conclusion and closing remarks

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KANSAS CITY

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Thank you!

LFSA REACH24 Organizing Committee

Suzanne MacFarland, M.D.

Kara Maxwell, M.D., Ph.D.

Judy Garber, M.D.

David Malkin, M.D.

Jenn Perry

Holly Fraumeni

Debbie Soprano